# Cataract as a phenotypic marker for a mutation in WFS1, the Wolfram syndrome gene



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| Titre | Cataract as a phenotypic marker for a mutation in WFS1, the Wolfram syndrome gene |
| Type de publication | Article de revue |
| Auteur | Titah, Salah Mohamed Cherif [1], Meunier, Isabelle [2], Blanchet, Catherine [3], Lopez, Severine [4], Rondouin, Gerard [5], Lenaers, Guy [6], Amati-Bonneau, Patrizia [7], Reynier, Pascal [8], Paquis-Flucklinger, Veronique [9], Hamel, Christian [10] |
| Pays | Italie |
| Editeur | Wichtig Editore |
| Ville | Milan |
| Type | Article scientifique dans une revue à comité de lecture |
| Année | 2012 |
| Langue | Anglais |
| Date | 2012 Mar-Apr |
| Numéro | 2 |
| Pagination | 254-258 |
| Volume | 22 |
| Titre de la revue | European Journal of Ophtalmology |
| ISSN | 1724-6016 |
| Mots-clés | Adult [11], Cataract [12], Cataract Extraction [13], DNA, Mitochondrial [14], Evoked Potentials, Visual [15], Exons [16], Female [17], Genetic Markers [18], GTP Phosphohydrolases [19], Heterozygote [20], Humans [21], Membrane Proteins [22], Mutation [23], Pedigree [24], Phenotype [25], proteins [26], Visual Acuity [27], Visual Fields [28], Wolfram Syndrome [29], Young Adult [30] |

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| Résumé en anglais | **PURPOSE:** Wolfram syndrome (WS) or diabetes insipidus, diabetes mellitus, optic atrophy, and deafness (DIDMOAD) (OMIM 222300) is an inherited neurodegenerative disease characterized by diabetes mellitus and optic atrophy as the 2 major criteria, followed later in life by deafness, diabetes insipidus, and various signs of neurologic impairment. The presence of a cataract has been variably mentioned in WS. **METHOD:** Two members of a family had thorough ophthalmic examination and their DNA was screened for mutations in mitochondrial DNA, WFS1, OPA1, and OPA3 genes. **RESULTS:** We report a patient who first had surgery for bilateral cataract at age 5 and who subsequently presented typical signs of WS, i.e., diabetes mellitus, optic atrophy with reduced visual acuity at 20/400 on both eyes at age 22, and mild deafness. The patient was found to be a compound heterozygote for 2 truncating mutations in WFS1, the major WS gene. She carried the previously reported c.1231_1233 delCT and a novel c.2431_2465dup35 mutation. She also was heterozygote for a novel OPA1 sequence variant, c.929A>G in exon 9, whose pathogenicity remains uncertain. The patient's mother was a heterozygous carrier of the c.2431_2465dup35 mutation. She did not have diabetes mellitus or optic atrophy but had bilateral polar cataract. She did not carry the OPA1 sequence variant. **CONCLUSIONS:** Cataract could be a marker for the WFS1 heterozygosity in this family, namely the c.2431_2465dup35 mutation. |
| URL de la notice | http://okina.univ-angers.fr/publications/ua9363 [31] |
| DOI | 10.5301/EJO.2011.8370 [32] |
| Lien vers le document | http://www.eur-j-ophthalmol.com/article/cataract-as-a-phenotypic-marker-... [33] |
| Titre abrégé | Eur J Ophthalmol |
| Identifiant (ID) PubMed | 21623591 [34] |

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